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NLP for Biomedical Applications

Information integration through terminology integration

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Introduction

- NLP and text mining require
  - Terminology
  - Domain knowledge
- Biomedical terminologies
  - Usually provide vocabulary
  - May provide some domain knowledge
  - Enable semantic integration
- Semantic integration may benefit NLP by enabling links to external resources

Terminology integration

The Unified Medical Language System

Unified Medical Language System

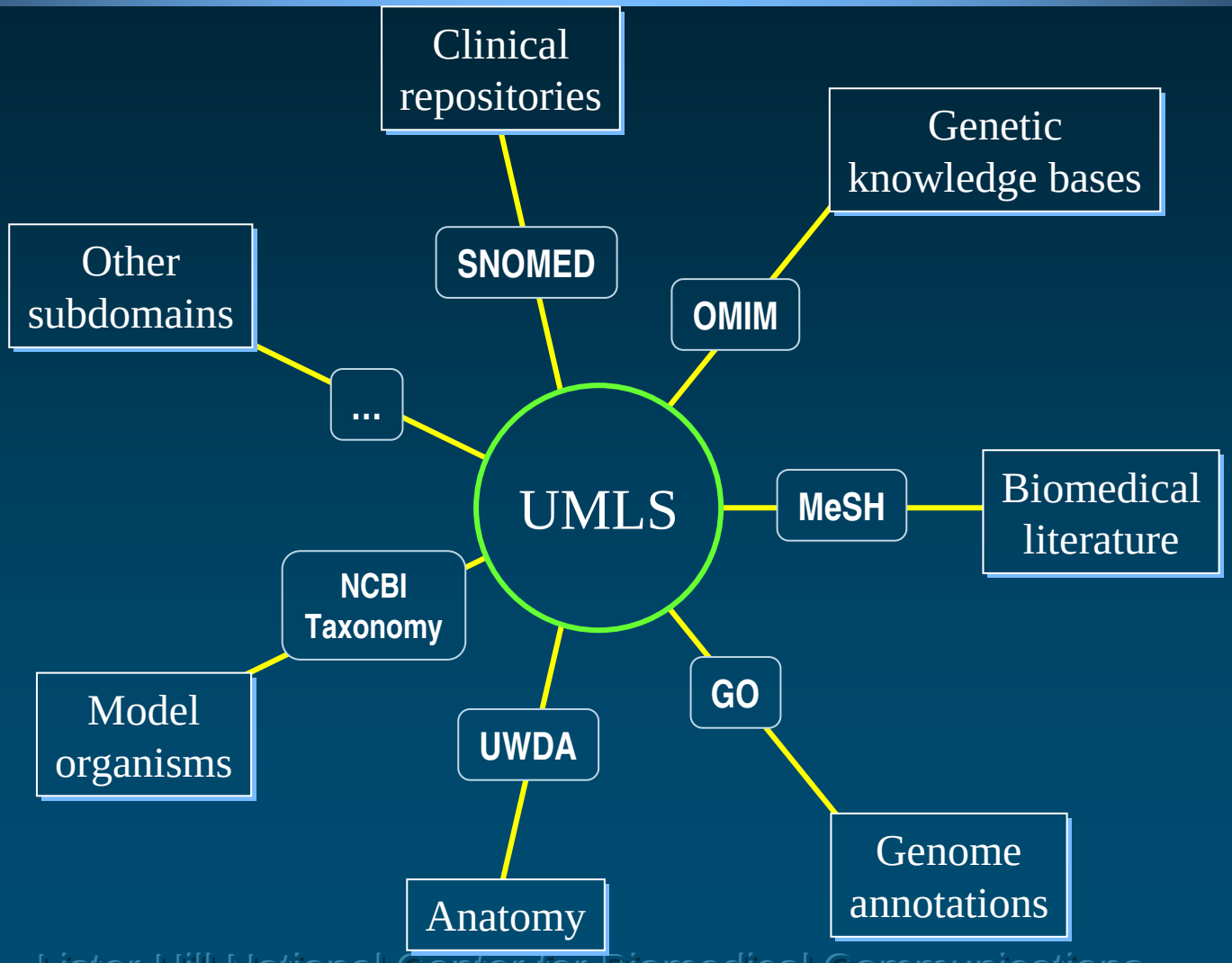
- Started in 1986
- National Library of Medicine

«[…] the UMLS project is an effort to overcome two significant barriers to effective retrieval of machine-readable information.

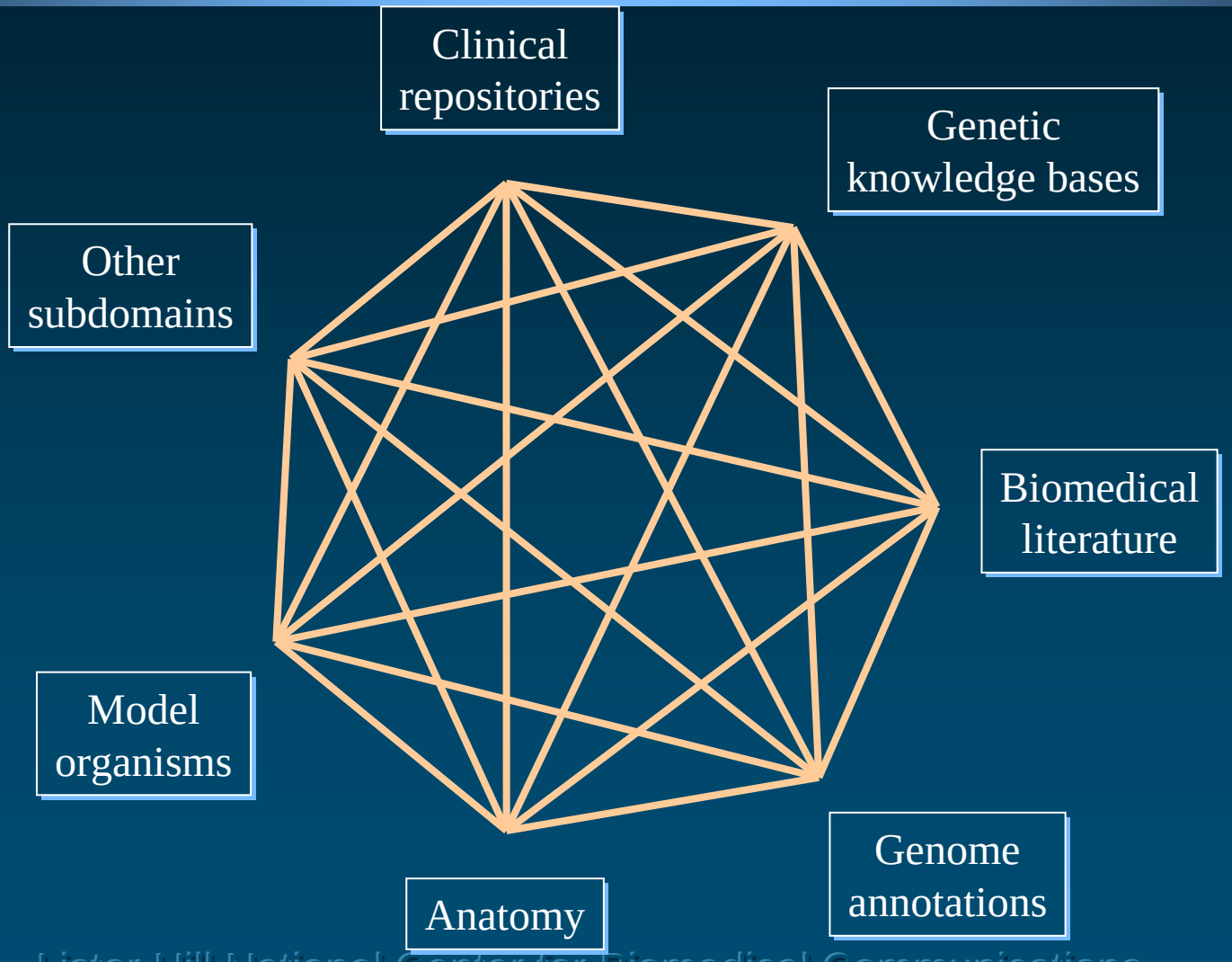
- The first is the variety of ways the same concepts are expressed in different machine-readable sources and by different people.
- The second is the distribution of useful information among many disparate databases and systems.»

- Terminology integration
  - 60 families of biomedical vocabularies

Integrating subdomains



Integrating subdomains



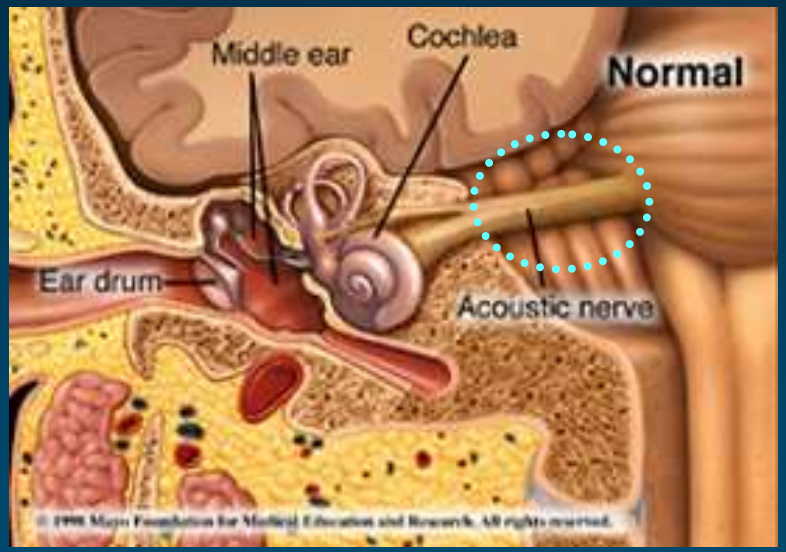
Information integration

Genetics as an example

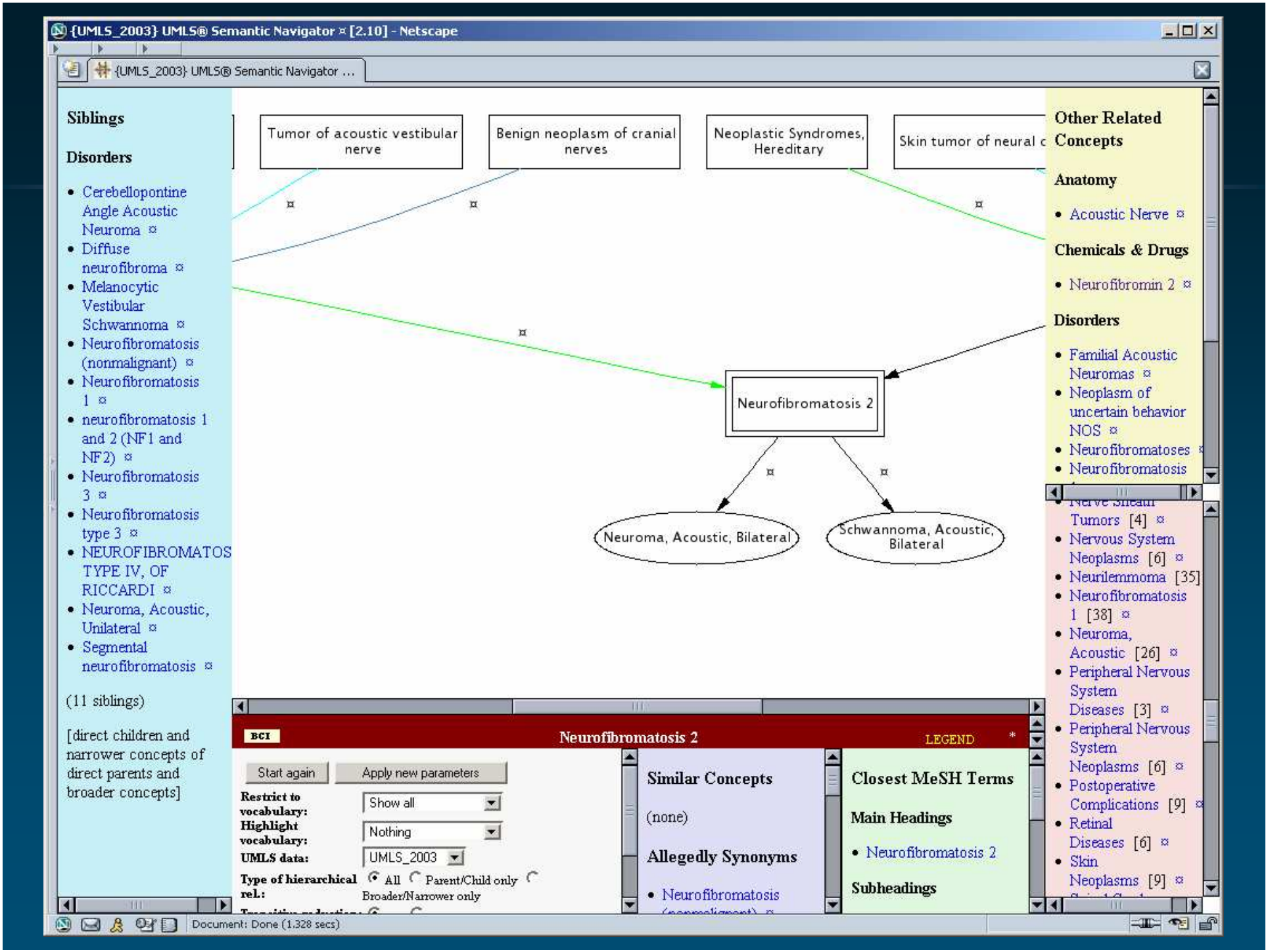
NF2 Gene, protein, and disease

Neurofibromatosis 2 is an autosomal dominant disease characterized by tumors called **schwannomas** involving the acoustic nerve, as well as other features. The disorder is caused by mutations of the **NF2** gene resulting in absence or inactivation of the protein product. The protein product of NF2 is commonly called **merlin** (but also **neurofibromin 2** and **schwannomin**) and functions as a tumor suppressor.

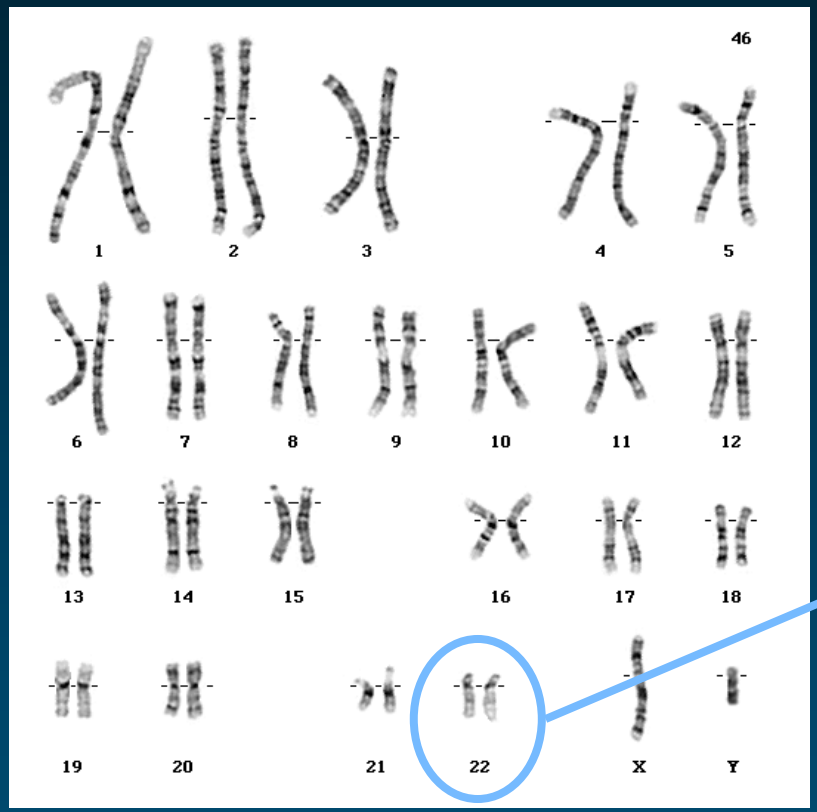
Schwannoma (acoustic neuroma)



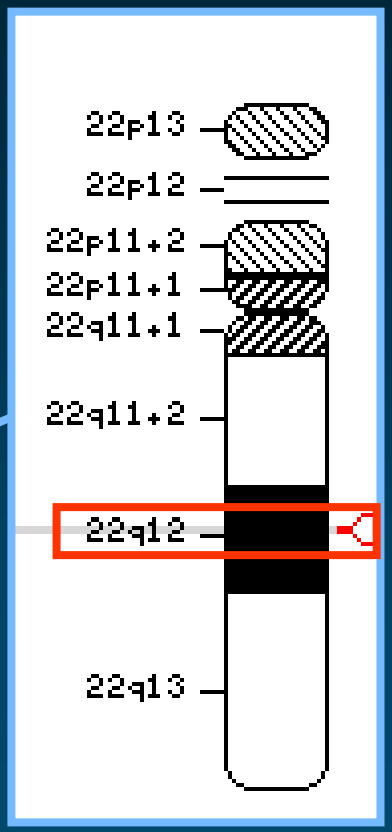
http://www.mayoclinic.com


{UMLS_2003} UMLS® Semantic Navigator ¤ [2.10] - Netscape
{UMLS_2003} UMLS® Semantic Navigator ...
Siblings
Disorders
Cerebellopontine Angle Acoustic Neuroma ¤
Diffuse neurofibroma ¤
Melanocytic Vestibular Schwannoma ¤
Neurofibromatosis (nonmalignant) ¤
Neurofibromatosis 1 ¤
neurofibromatosis 1 and 2 (NF1 and NF2) ¤
Neurofibromatosis 3 ¤
Neurofibromatosis type 3 ¤
NEUROFIBROMATOSIS TYPE IV, OF RICCARDI ¤
Neuroma, Acoustic, Unilateral ¤
Segmental neurofibromatosis ¤
(11 siblings)
[direct children and narrower concepts of direct parents and broader concepts]
Tumor of acoustic vestibular nerve
Benign neoplasm of cranial nerves
Neoplastic Syndromes, Hereditary
Skin tumor of neural c
Neurofibromatosis 2
Neuroma, Acoustic, Bilateral
Schwannoma, Acoustic, Bilateral
Other Related Concepts
Anatomy
Acoustic Nerve ¤
Chemicals & Drugs
Neurofibromin 2 ¤
Disorders
Familial Acoustic Neuromas ¤
Neoplasm of uncertain behavior NOS ¤
Neurofibromatoses ¤
Neurofibromatosis
Tumors [4] ¤
Nervous System Neoplasms [6] ¤
Neurilemmoma [35]
Neurofibromatosis 1 [38] ¤
Neuroma, Acoustic [26] ¤
Peripheral Nervous System Diseases [3] ¤
Peripheral Nervous System Neoplasms [6] ¤
Postoperative Complications [9] ¤
Retinal Diseases [6] ¤
Skin Neoplasms [9] ¤
BCI
Neurofibromatosis 2
LEGEND
Start again
Apply new parameters
Restrict to vocabulary:
Show all
Highlight vocabulary:
Nothing
UMLS data:
UMLS_2003
Type of hierarchical rel.:
All
Parent/Child only
Broader/Narrower only
Similar Concepts
(none)
Allegedly Synonyms
Neurofibromatosis
Closest MeSH Terms
Main Headings
Neurofibromatosis 2
Subheadings
Document: Done (1.328 secs)

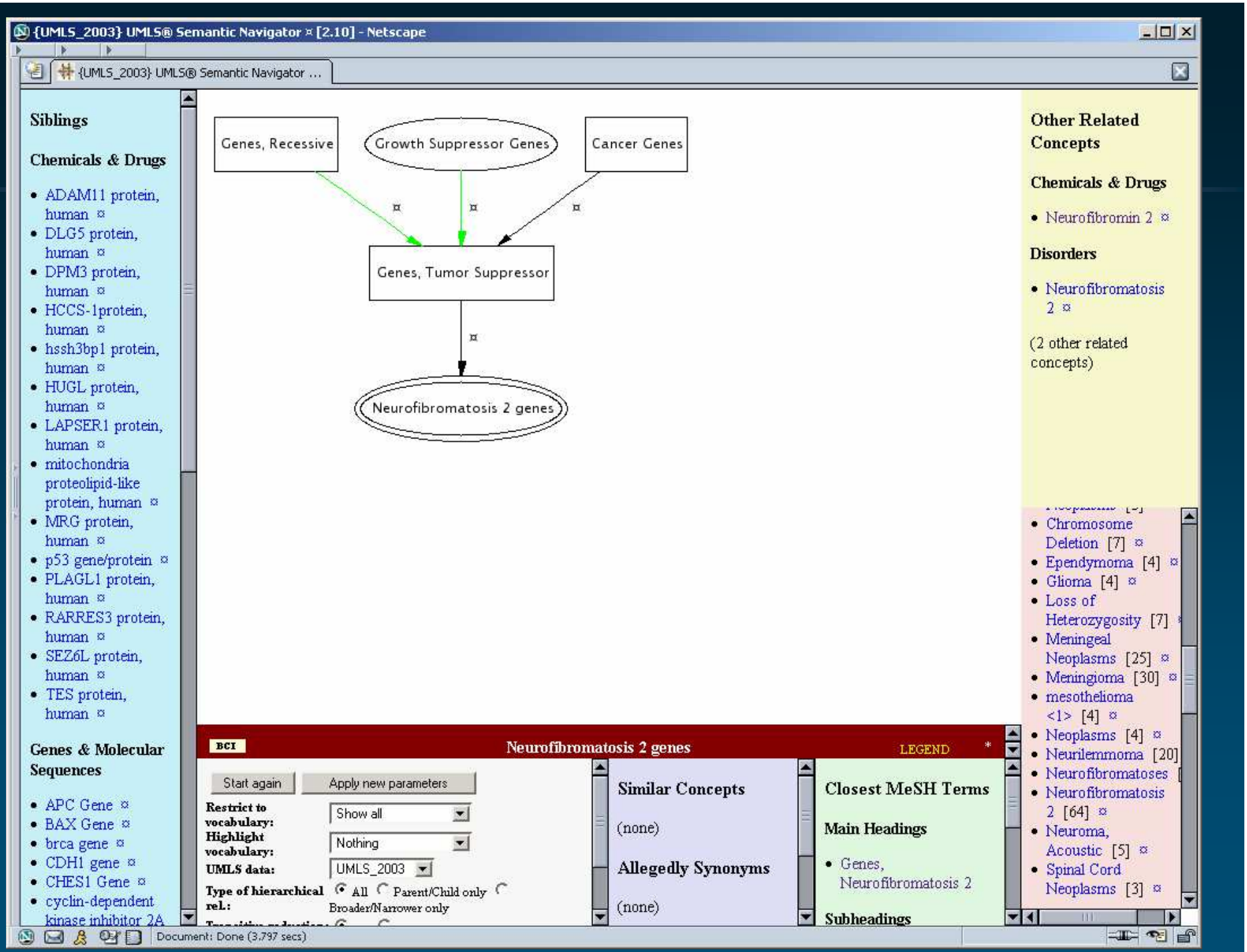
NF2 gene



http://staff.washington.edu/timk/cyto/human/

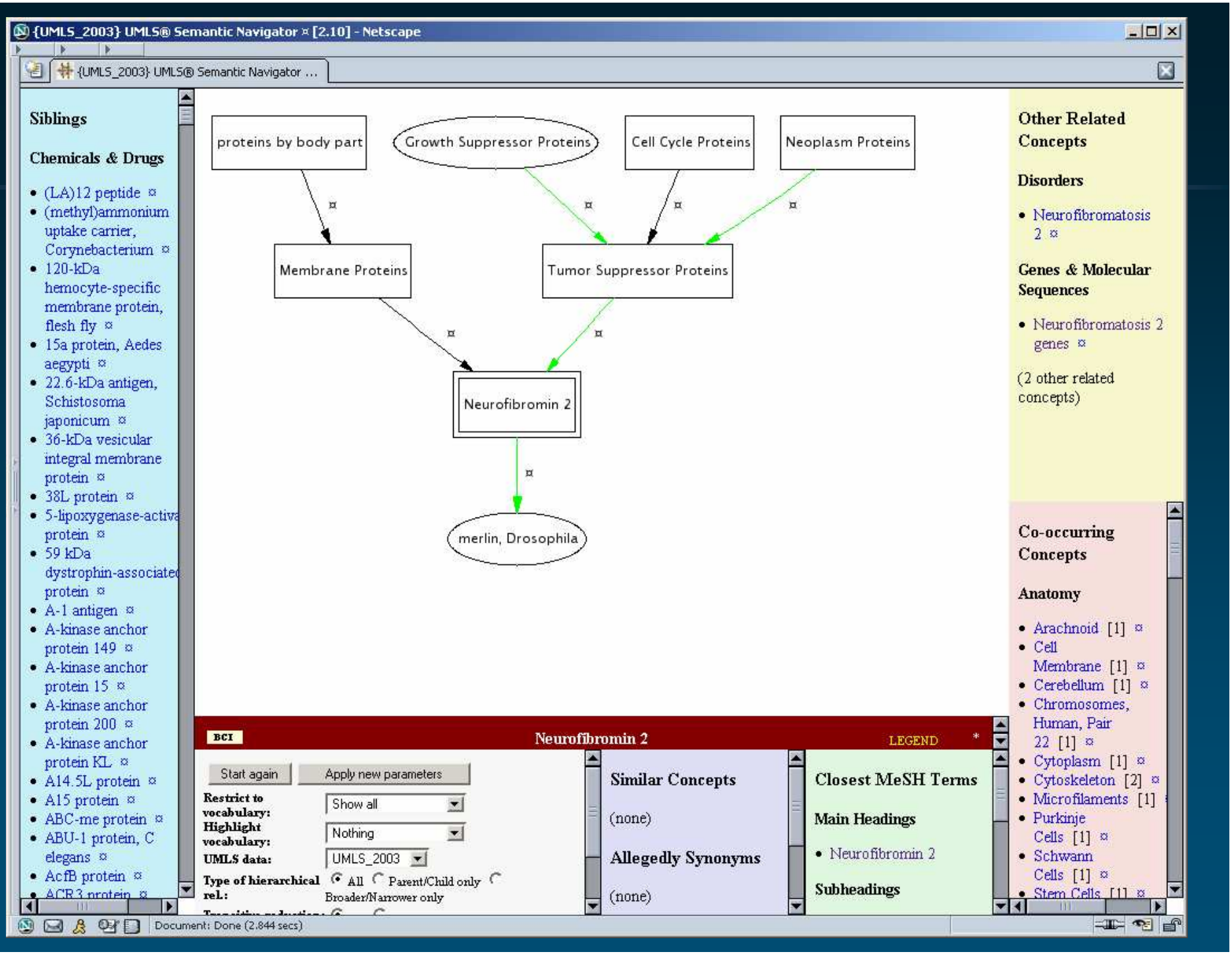


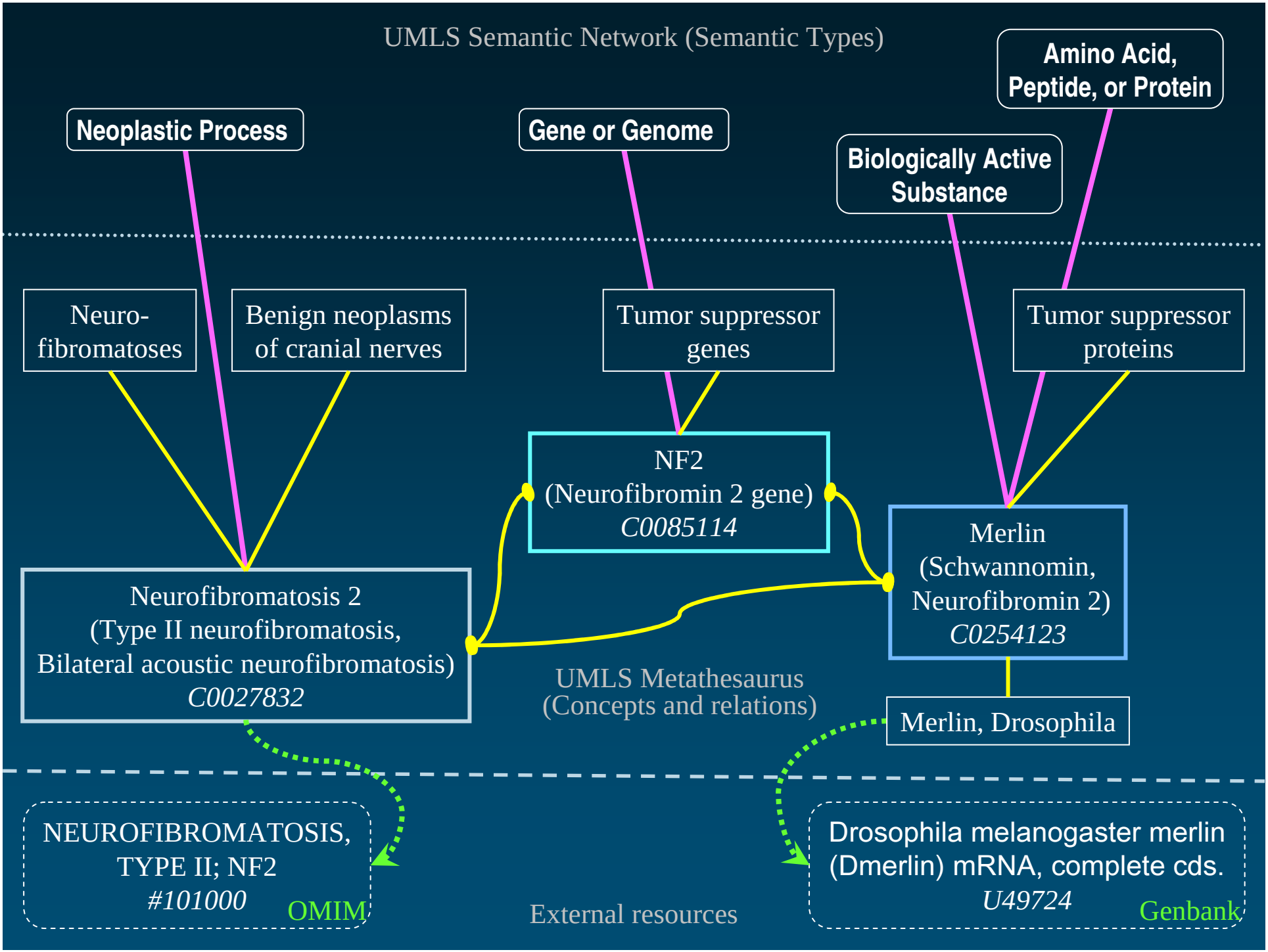
http://www.ncbi.nlm.nih.gov/mapview/


{UMLS_2003} UMLS® Semantic Navigator ¤ [2.10] - Netscape
{UMLS_2003} UMLS® Semantic Navigator ...
Siblings
Chemicals & Drugs
ADAM11 protein, human ¤
DLG5 protein, human ¤
DPM3 protein, human ¤
HCCS-1protein, human ¤
hssh3bp1 protein, human ¤
HUGL protein, human ¤
LAPSER1 protein, human ¤
mitochondria proteolipid-like protein, human ¤
MRG protein, human ¤
p53 gene/protein ¤
PLAGL1 protein, human ¤
RARRES3 protein, human ¤
SEZ6L protein, human ¤
TES protein, human ¤
Genes & Molecular Sequences
APC Gene ¤
BAX Gene ¤
brca gene ¤
CDH1 gene ¤
CHES1 Gene ¤
cyclin-dependent kinase inhibitor 2A
Genes, Recessive
Growth Suppressor Genes
Cancer Genes
Genes, Tumor Suppressor
Neurofibromatosis 2 genes
Other Related Concepts
Chemicals & Drugs
Neurofibromin 2 ¤
Disorders
Neurofibromatosis 2 ¤
(2 other related concepts)
Chromosome Deletion [7] ¤
Ependymoma [4] ¤
Glioma [4] ¤
Loss of Heterozygosity [7] ¤
Meningeal Neoplasms [25] ¤
Meningioma [30] ¤
mesothelioma <1> [4] ¤
Neoplasms [4] ¤
Neurilemmoma [20]
Neurofibromatoses
Neurofibromatosis 2 [64] ¤
Neuroma, Acoustic [5] ¤
Spinal Cord Neoplasms [3] ¤
BCI
Neurofibromatosis 2 genes
LEGEND
Start again
Apply new parameters
Restrict to vocabulary:
Show all
Highlight vocabulary:
Nothing
UMLS data:
UMLS_2003
Type of hierarchical rel.:
All
Parent/Child only
Broader/Narrower only
Similar Concepts
(none)
Allegedly Synonyms
(none)
Closest MeSH Terms
Main Headings
Genes, Neurofibromatosis 2
Subheadings
Document: Done (3.797 secs)

Merlin

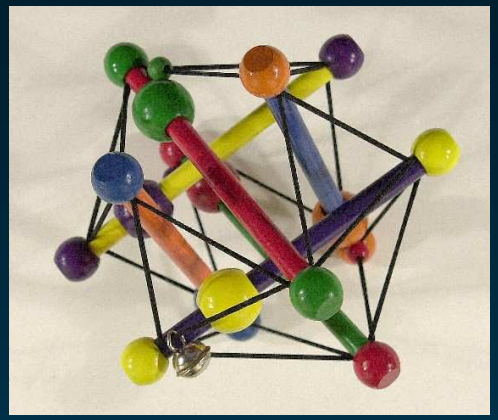
- Synonyms
  - Neurofibromin 2
  - Schwannomin
  - Schwannomerlin
  - Neurofibromatosis-2
- 10 isoforms
- Annotations
  - Negative regulation of cell proliferation
  - Cytoskeleton
  - Plasma membrane


{UMLS_2003} UMLS® Semantic Navigator ¤ [2.10] - Netscape
{UMLS_2003} UMLS® Semantic Navigator ...
Siblings
Chemicals & Drugs
(LA)12 peptide ¤
(methyl)ammonium uptake carrier, Corynebacterium ¤
120-kDa hemocyte-specific membrane protein, flesh fly ¤
15a protein, Aedes aegypti ¤
22.6-kDa antigen, Schistosoma japonicum ¤
36-kDa vesicular integral membrane protein ¤
38L protein ¤
5-lipoxygenase-activa protein ¤
59 kDa dystrophin-associated protein ¤
A-1 antigen ¤
A-kinase anchor protein 149 ¤
A-kinase anchor protein 15 ¤
A-kinase anchor protein 200 ¤
A-kinase anchor protein KL ¤
A14.5L protein ¤
A15 protein ¤
ABC-me protein ¤
ABU-1 protein, C elegans ¤
AcfB protein ¤
ACR3 protein ¤
proteins by body part
Growth Suppressor Proteins
Cell Cycle Proteins
Neoplasm Proteins
Membrane Proteins
Tumor Suppressor Proteins
Neurofibromin 2
merlin, Drosophila
Other Related Concepts
Disorders
Neurofibromatosis 2 ¤
Genes & Molecular Sequences
Neurofibromatosis 2 genes ¤
(2 other related concepts)
Co-occurring Concepts
Anatomy
Arachnoid [1] ¤
Cell Membrane [1] ¤
Cerebellum [1] ¤
Chromosomes, Human, Pair 22 [1] ¤
Cytoplasm [1] ¤
Cytoskeleton [2] ¤
Microfilaments [1] ¤
Purkinje Cells [1] ¤
Schwann Cells [1] ¤
Stem Cells [1] ¤
BCI
Neurofibromin 2
LEGEND *
Start again
Apply new parameters
Restrict to vocabulary:
Show all
Highlight vocabulary:
Nothing
UMLS data:
UMLS_2003
Type of hierarchical rel.:
All
Parent/Child only
Broader/Narrower only
Similar Concepts
(none)
Allegedly Synonyms
(none)
Closest MeSH Terms
Main Headings
Neurofibromin 2
Subheadings
Document: Done (2.844 secs)


UMLS Semantic Network (Semantic Types)
Neoplastic Process
Gene or Genome
Biologically Active Substance
Amino Acid, Peptide, or Protein
Neuro-fibromatoses
Benign neoplasms of cranial nerves
Tumor suppressor genes
Tumor suppressor proteins
NF2 (Neurofibromin 2 gene) C0085114
Merlin (Schwannomin, Neurofibromin 2) C0254123
Neurofibromatosis 2 (Type II neurofibromatosis, Bilateral acoustic neurofibromatosis) C0027832
UMLS Metathesaurus (Concepts and relations)
Merlin, Drosophila
NEUROFIBROMATOSIS, TYPE II; NF2 #101000
OMIM
External resources
Drosophila melanogaster merlin (Dmerlin) mRNA, complete cds. U49724
Genbank

Limitations

- Genes not systematically represented
  - Most gene products and diseases are
- Gene/Gene product-Disease relations
  - Not systematically represented
  - Not explicitly represented (e.g., co-occurrence)
- Cross-references not systematically represented
- Naming conventions (genes)



Medical Ontology Research

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